Phenylketonuria (PKU)
General Overview

Q. What is PKU?

A. PKU is a disease that affects the way the body processes protein. Children with PKU cannot process a part of the protein called phenylalanine. As a result, phenylalanine builds up in the bloodstream and causes brain damage.

Q. How does the body normally process phenylalanine?

A. The body normally converts phenylalanine into tyrosine which is in turn used in other functions. This conversion is made possible by the presence of an enzyme called phenylalanine hydroxylase.

Q. What happens to phenylalanine in a child with PKU?

A. A child with PKU has an absence of the enzyme (phenylalanine hydroxylase) needed to convert phenylalanine to tyrosine. Failure of the conversion to take place results in a buildup of phenylalanine. Through a mechanism that is not well understood, the excess phenylalanine is toxic to the central nervous system.

Q. What are the effects of having PKU?

A. Untreated PKU results in severe mental retardation.

Q. Is there only one form of PKU?

A. Yes, but not every child has the same degree of enzyme deficiency so the level of treatment may vary.

Q. What is Maternal PKU?

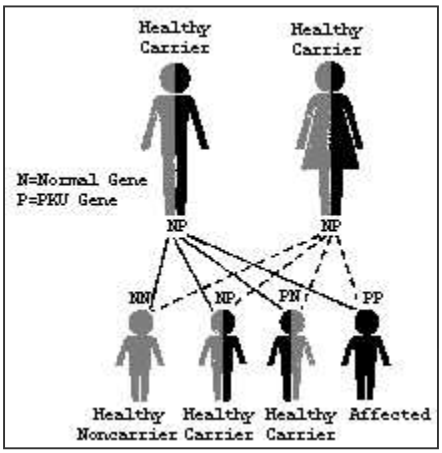
A. Maternal PKU is a concern for women with PKU. These women have high levels of phenylalanine in their blood and have a very large probability of harming their unborn baby. Most of these babies do not inherit PKU, but suffer from brain damaged cause by the mother's high phenylalanine levels before birth. Maternal PKU has become a significant concern as young women who were once taken off the diet are now reaching childbearing ages in ever-increasing numbers. A large collaborative study has shown that returning the woman to diet before conception and keeping blood phenylalanine levels below 6 mg/dl results in the best outcome for the baby. Many children born to mothers with PKU are developing normally because of early and strict treatment.

Q. How common is PKU?

A. PKU occurs about once out of 16,000 births in the United States and is distributed equally between the sexes.

Q. How does a child get PKU?

A. PKU is inherited when both parents have the PKU gene and both pass it on to their baby. A parent who has the PKU gene, but not the disease, is called a "carrier." A carrier has a normal gene as well as a PKU gene in each cell. A carrier's health is not affected in any known way.



When both parents are carriers, there is a one-in-four chance that both will pass the PKU gene on to a child, causing the child to be born with the disease. There also is a one-in-four chance that they will each pass on a normal gene, and the child will be free of the disease. There is a two-in-four chance that a baby will inherit the PKU gene from one parent and the normal gene from the other, making it a carrier like its parents. These chances are the same in each pregnancy.

Q. What is the treatment for PKU?

A. Mental retardation caused by PKU can be totally prevented if the baby is treated with a special diet that is low in phenylalanine begun in the first three weeks of life.
At first, the baby is fed a special formula that contains protein but no phenylalanine. Breast milk or infant formula is used sparingly to supply as much phenylalanine as the baby can tolerate. Later, certain vegetables, fruits, some grain products (for example, certain cereals and noodles) and other low-phenylalanine foods are added to the diet, but no regular milk, cheese, eggs, meat, fish and other high protein foods are ever allowed. Since protein is essential for normal growth and development, the child must continue to have one of the special formulas which are high in protein and essential nutrients, but contain little or no phenylalanine. Diet drinks and foods that contain the artificial sweetener aspartame (sold as Nutrasweet or Equal) must be strictly avoided. Children and adults with PKU require follow-up care at a medical center or clinic that specializes in this disorder. The diet for each person must be individualized, depending on how much phenylalanine they can tolerate, his or her age, weight and other factors. All affected persons need regular blood tests to measure if the levels of phenylalanine are too high or too low. The diet must then be adjusted accordingly.
Individuals with PKU must remain on a restricted diet throughout childhood and adolescence, and perhaps for life. Until the 1980s, health care providers believed that children with PKU could safely discontinue their special diet around age 6 when brain growth was completed. However, high blood levels of phenylalanine in children and adolescents can lead to a decrease in IQ, to learning disabilities, and to behavioral disturbances in most -- but perhaps not all -- children with PKU.
There currently is no good way to predict which individuals with PKU could discontinue their diets without adverse effects. Parents of children with PKU and affected adults should discuss their diet and treatment questions with health care professionals at one of the special clinics for PKU.

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